NINDS funded Research Meeting on Sturge-Weber syndrome

“Leveraging a Gene Discovery; an Agenda for Future Research”

Date: April 19-20, 2015
Place: Bethesda Marriot Suites, Bethesda Maryland

Organizing Committee
Chair: Anne M Comi M.D., Dept. of Neurology, Kennedy Krieger Institute
Karen L. Ball, Sturge-Weber Foundation President and CEO
Mustafa Sahin, M.D., Ph.D., Dept. of Neurology, Boston Children’s Hospital, Harvard Medical School
Alex V. Levin, M.D., MHSc, Pediatric Ophthalmology and Ocular Genetics, Wills Eye Hospital
Michael Gold, M.D., Department of Dermatology, Vanderbilt University
Charles Swindell, Ph.D., Volunteer Advisor
NIH Program Director: Jill Morris, Ph.D. Neurogenetics, NINDS

The discovery of the causative somatic mutation in GNAQ published in 2013 has transformed the research possibilities for Sturge-Weber syndrome. Yet, many questions about the manifestations of this syndrome remain unanswered. At this exciting time, the National Institutes of Health is spearheading efforts to focus future research goals and milestones to hasten translation towards novel and effective treatments for Sturge-Weber syndrome. We invite researchers, both basic and clinical to participate in this workshop, which will feature leaders in the field and aims to foster new collaborations. The workshop will bring forward young investigators, and also seeks to attract new researchers into Sturge-Weber syndrome research.

Scheduled speakers, presenters and discussion moderators include the following:

Anne M. Comi, M.D. Associate Professor Neurology and Pediatrics, Kennedy Krieger Institute and Johns Hopkins School of Medicine. Dr. Comi is internationally known for her clinical and translational research in Sturge-Weber syndrome (SWS). She directs a Sturge-Weber syndrome Clinical Center and a lab focused on SWS, and oversees multi-centered SWS clinical research funded through the Brain Vascular Malformation Consortium.

Karen L. Ball President and CEO of the Sturge-Weber Foundation and advocate for Sturge-Weber syndrome awareness and research for the past 25 years. The Sturge-Weber Foundation is an international leader in working with clinicians, researchers and families to develop new clinical and research programs to better understand and treat Sturge-Weber syndrome.

NEUROLOGY

Mustafa Sahin, M.D., Ph.D. Associate Professor of Neurology, Boston Children’s Hospital, Harvard Medical School. Dr. Sahin is internationally recognized for his research concerning the TSC/mTOR pathway and his laboratory is directed at understanding the cellular mechanisms of neuronal connectivity and its relationship to neurological dysfunction. He also directs a Center for Sturge-Weber syndrome and is a medical director for the Sturge-Weber Foundation.

Csaba Juhasz, M.D., Ph.D. Professor of Pediatrics and Neurology, Wayne State University School of Medicine, Children’s Hospital of Michigan. Dr. Juhasz is an international expert in functional neuroimaging. His research has provided crucial insights into the evolution of Sturge-Weber syndrome brain involvement and neurologic impairments.
OPHTHALMOLOGY
Alex V. Levin, M.D., MHSc Professor of Ophthalmology and Pediatrics, Sydney Kimmel Medical College of Thomas Jefferson University. Dr. Levin is Chief of the Pediatric Ophthalmology and Ocular Genetics service at Wills Eye Hospital and an expert in glaucoma management in Sturge-Weber syndrome. He participates in multi-centered SWS clinical research through the Brain Vascular Malformation Consortium and other related projects. He is the Chair of the Medical Advisory Board of the Sturge-Weber Foundation.
Sharon Freedman, M.D. Professor of Ophthalmology and Pediatrics, Duke University. Dr. Freedman is a clinician-educator with specific expertise and interest in the area of pediatric glaucoma and pediatric oculars disorders associated with glaucoma including Sturge-Weber syndrome.
Timothy Murray, M.D., MBA, FACS Founding Director of Murray Ocular Oncology and Retina. Dr. Murray brings longstanding clinical expertise and research interest in both Sturge-Weber syndrome and uveal melanoma both of which have been demonstrated to be caused by/associated with the same somatic mutation, though presenting in distinctly different cell types.

DERMATOLOGY
Michael H. Gold, M.D., Clinical Instructor in Dermatology, Vanderbilt University Medical Center; Owner of Gold Skin Care Center and of Tennessee Clinical Research Center. Dr. Gold is an internationally recognized expert in laser treatment of vascular birthmarks and for providing patients with leading edge technological advances.
Adelaide A. Hebert, M.D. Professor of Dermatology and Pediatrics, University of Texas Health Science Center at Houston. Dr. Hebert has completed important clinical drug trials for many dermatologic conditions including multi-centered, placebo-controlled, and pharmaceutical company sponsored trials.

ENDOCRINOLOGY
Emily Germain-Lee, M.D. Associate Professor Pediatric Endocrinology, Kennedy Krieger Institute and Johns Hopkins School of Medicine. Dr. Germain-Lee is a clinician scientist with expertise in the endocrinologic aspects of SWS and in the pathobiology of G-proteins in another rare disorder, Albright hereditary osteodystrophy.

VASCULAR BIOLOGY AND PATHOGENESIS
Paula North, M.D., Ph. D. Professor of Pathology, Medical College of Wisconsin. Dr. North is internationally recognized for her expertise in vascular pathology and her many important research contributions to the field of vascular biology.
Doug Marchuk, Ph. D. James B. Duke Professor of Molecular Genetics and Microbiology, Duke University. Dr. Marchuk is internationally known for his contributions to the genetics and molecular biology of several vascular malformations, including Cerebral Cavernous Malformations, Hereditary Hemorrhagic Telangiectasia and Sturge-Weber syndrome. He is Co-PI of the Brain Vascular Malformation Consortium.
Nancy Ratner, M.D., Ph. D. Professor, Experimental Hematology and Cancer Biology, Cincinnati Children’s Hospital. Dr. Ratner’s expertise is in G-protein pathways and related conditions, including Neurofibromatosis type 1.
Adrienne Hammill, M.D., Ph.D. Assistant Professor Dept. of Hematology and Oncology, Cincinnati Children’s Hospital Medical Center. Dr. Hammill’s research has involved the treatment of lymphatic and venous malformations with mTOR inhibitors and other agents. She is involved with Sturge-Weber syndrome multi-centered clinical research through the Brain Vascular Malformation Consortium.

POSTER SESSION
Meeting will include a Poster Session. All abstracts must be submitted to by Friday February 15th, 2015. Abstracts will be reviewed and submitters informed of results by February 25th, 2015. Travel funds are available for Young Investigators and Trainees who are encouraged to submit abstracts and to attend the meeting.

Registration and Hotel Reservations
Registrations will be first come, first serve. Registrations can be made by emailing Dr. Anne Comi at comi@kennedykrieger.org or calling 443-923-9569. To make room reservations at the reduced meeting rate call the Bethesda Marriott Suites and request the SW: Leveraging Gene Discovery room rate by March 16th, 2015.